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STAT FISH Chromosome 18

Test Code: CF18S
Turnaround time: 1 day - 3 days (All abnormal findings are called out immediately.)
CPT Codes: 88271 x2, 88275 x2, 88291 x1

Condition Description

Chromosome disorders due to non-disjunction of chromosomes 13, 18, 21, X and Y together comprise the majority of the microscopically detectable chromosome disorders. Aneuploidy of the non-sex determining chromosomes increases in frequency along with increasing maternal age.

Analysis by Fluorescence *In Situ* Hybridization (FISH) allows for the most rapid detection of the most common chromosome disorders. Results can typically be reported in 24-48 hours from the time of receipt.

Concurrent G-banded chromosome analysis or chromosomal microarray is required.

Trisomy 18 or Edward syndrome is a disorder characterized by low birth weight, heart defects, and gastrointestinal malformations. Trisomy 18 occurs in 1 out of every 5,000 newborns. Advanced maternal age increases the risk for an infant with trisomy 18. Individuals with trisomy 18 have severe intellectual disabilities. Physical characteristics may include clenched hands with overlapping fingers, rocker bottom feet and microcephaly. The median survival time is 14.5 days. Five to 10% of children with trisomy 18 live past year one of life; however, those that do will have severe anomalies which usually leads to their death. Ultrasound findings for trisomy 18 include small for gestational age (IUGR), strawberry shaped skull, choroid plexus cysts, cardiac and brain abnormalities. Approximately 80% of newborns with trisomy 18 are female.

Most cases of trisomy 18 result from an extra copy of chromosome 18. This extra genetic material disrupts the developmental process and causes the characteristic features of trisomy 18. In individuals with a mosaic form of trisomy 18, the severity can range from normal to severely affected. Trisomy 18 is not typically inherited, but rather a random event of nondisjunction in meiosis. Trisomy 18 caused by a translocation can be inherited and translocation carriers are at risk of having a child with trisomy 18. Approximately 20% of cases are caused by a translocation.

References:

1. www.ghr.nlm.nih.gov/condition/trisomy-18
2. Smith, D.W. (2006) Trisomy 18 Syndrome. Smith's Recognizable Patterns of Human Malformation. Philadelphia, Elsevier Saunders, 6th Edit. pp. 13-17.

Indications

- Multiple congenital anomalies
- Dysmorphic features
- IUGR
- Advanced maternal age (AMA)
- Abnormal ultrasound
- Abnormal serum screen

Methodology

Interphase FISH analysis is performed on uncultured peripheral blood samples using commercially available probes.

Detection

FISH is very sensitive in the detection of aneuploidy. This probe set is specific to chromosome 18 and only numerical abnormalities of chromosome 18 will be detected. Validation for specificity and sensitivity performed on each probe. Control probes are present in all probe sets.

Specimen Requirements

Type: Whole Blood (EDTA and Sodium Heparin)

Specimen Requirements:
Sodium Heparin and EDTA
Infants (Children (>2 years): 3-5 ml in both tubes
Older Children & Adults: 7-10 ml in both tubes

Specimen Collection and Shipping:
Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

Special Instructions

Concurrent G-banded chromosome analysis or chromosomal microarray is required.

Related Tests

- Chromosomal Microarray, EmArray Cyto (VA)

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- Chromosome Analysis (CA/CB)